Cardiac Phenotypes in Hereditary Muscle Disorders: JACC State-of-the-Art Review.

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Abstract
Hereditary muscular diseases commonly involve the heart. Cardiac manifestations encompass a spectrum of phenotypes, including both cardiomyopathies and rhythm disorders. Common biomarkers suggesting cardiomyocutaneous diseases include increased circulating creatine kinase and/or lactic acid levels or disease-specific metabolic indicators. Cardiac and extra-cardiac traits, imaging tests, family studies, and genetic testing provide precise diagnoses. Cardiac phenotypes are mainly dilated and hypokinetically in dystrophinopathies, Emery-Dreifuss muscular dystrophies, and limb girdle muscular dystrophies; hypertrophic in Friedreich ataxia, mitochondrial diseases, glycogen storage diseases, and fatty acid oxidation disorders; and restrictive in myofibrillar myopathies. Left ventricular noncompaction is variably associated with the different myopathies. Conduction defects and arrhythmias constitute a major phenotype in myotonic dystrophies and skeletal muscle channelopathies. Although the actual cardiac management is rarely based on the cause, the cardiac phenotypes need precise characterization because they are often the only or the predominant manifestations and the prognostic determinants of many hereditary muscle disorders.

KEYWORDS: arrhythmias; cardiomyopathies; dystrophy; myopathy; skeletal muscle

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